oculofaciocardiodental syndrome

Oculofaciocardiodental (OFCD) syndrome is a condition that affects the development of the eyes (oculo-), facial features (facio-), heart (cardio-) and teeth (dental). This condition occurs only in females.

The eye abnormalities associated with OFCD syndrome can affect one or both eyes. Many people with this condition are born with eyeballs that are abnormally small (microphthalmia). Other eye problems can include clouding of the lens (cataract) and a higher risk of glaucoma, an eye disease that increases the pressure in the eye. These abnormalities can lead to vision loss or blindness.

People with OFCD syndrome often have a long, narrow face with distinctive facial features, including deep-set eyes and a broad nasal tip that is divided by a cleft. Some affected people have an opening in the roof of the mouth called a cleft palate.

Heart defects are another common feature of OFCD syndrome. Babies with this condition may be born with a hole between two chambers of the heart (an atrial or ventricular septal defect) or a leak in one of the valves that controls blood flow through the heart (mitral valve prolapse).

Teeth with very large roots (radiculomegaly) are characteristic of OFCD syndrome. Additional dental abnormalities can include delayed loss of primary (baby) teeth, missing or abnormally small teeth, misaligned teeth, and defective tooth enamel.

Frequency

OFCD syndrome is very rare; the incidence is estimated to be less than 1 in 1 million people.

Genetic Changes

Mutations in the *BCOR* gene cause OFCD syndrome. The *BCOR* gene provides instructions for making a protein called the BCL6 corepressor. This protein helps regulate the activity of other genes. Little is known about the protein's function, although it appears to play an important role in early embryonic development. Several mutations in the *BCOR* gene have been found in people with OFCD syndrome. These mutations prevent the production of any functional protein from the altered gene, which disrupts the normal development of the eyes and several other organs and tissues before birth.

Inheritance Pattern

This condition is inherited in an X-linked dominant pattern. The gene associated with this condition is located on the X chromosome, which is one of the two sex

chromosomes. In females (who have two X chromosomes), a mutation in one of the two copies of the gene in each cell is sufficient to cause the disorder. Some cells produce a normal amount of BCL6 corepressor protein and other cells produce none. The resulting overall reduction in the amount of this protein leads to the signs and symptoms of OFCD syndrome.

In males (who have only one X chromosome), mutations result in a total loss of the BCL6 corepressor protein. A lack of this protein appears to be lethal very early in development, so no males are born with OFCD syndrome.

Other Names for This Condition

- MCOPS2
- Microphthalmia, cataracts, radiculomegaly, and septal heart defects
- Microphthalmia, syndromic 2
- Oculo-facio-cardio-dental syndrome
- OFCD syndrome

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Oculofaciocardiodental syndrome
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C1846265/

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Health Topic: Congenital Heart Defects
  https://medlineplus.gov/congenitalheartdefects.html
- Health Topic: Eye Diseases
  https://medlineplus.gov/eyediseases.html

Genetic and Rare Diseases Information Center

- Oculofaciocardiodental syndrome
  https://rarediseases.info.nih.gov/diseases/4628/oculofaciocardiodental-syndrome

Additional NIH Resources

- National Eye Institute: Facts About Anophthalmia and Microphthalmia
  https://nei.nih.gov/health/anoph/
- National Heart, Lung, and Blood Institute: Mitral Valve Prolapse
  https://www.nhlbi.nih.gov/health/health-topics/topics/mvp/

Educational Resources

- Genomics in Action: Leslie G. Biesecker, M.D.
  https://www.genome.gov/14514560/
- MalaCards: microphthalmia, syndromic 2
  http://www.malacards.org/card/microphthalmia_syndromic_2
- Orphanet: Oculofaciocardiodental syndrome
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=2712

Patient Support and Advocacy Resources

- American Foundation for the Blind
  http://www.afb.org/
- Resource list from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/anopthal.html

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22Eye+Diseases%2C+Hereditary%22+OR+%22oculofaciocardiodental+syndrome%22

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28oculofaciocardiodental+syndrome%5BTIAB%5D%29+OR+%28mcops2%5BTIAB%5D%29+OR+%28ofcd+syndrome%5BTIAB%5D%29+OR+%28oculo-facio-cardio-dental+syndrome%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- MICROPHTHALMIA, SYNDROMIC 2
  http://omim.org/entry/300166

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